



ATG16L1 gene

autophagy related 16 like 1

Normal Function

The *ATG16L1* gene provides instructions for making a protein called autophagy related 16-like 1. This protein is part of a larger family of proteins that are required for a process called autophagy. Cells use this process to recycle worn-out cell parts and break down certain proteins when they are no longer needed. Autophagy also plays an important role in controlled cell death (apoptosis). Additionally, autophagy is involved in the body's inflammatory response and helps the immune system destroy some types of harmful bacteria and viruses.

Health Conditions Related to Genetic Changes

Crohn disease

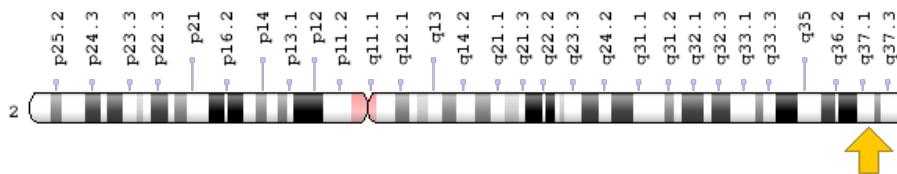
At least one variation in the *ATG16L1* gene is associated with an increased risk of Crohn disease, particularly a form of the disorder that affects the lower part of the small intestine (the ileum). This increased risk has been found primarily in white populations. The identified *ATG16L1* variation changes a single protein building block (amino acid) in a critical region of the autophagy related 16-like 1 protein. Specifically, it replaces the amino acid threonine with the amino acid alanine at protein position 300 (written as Thr300Ala or T300A).

The effects of variations in the *ATG16L1* gene on Crohn disease risk are unclear. Changes in this gene may affect the autophagy process, allowing worn-out cell parts and harmful bacteria to persist when they would otherwise be destroyed. These cell components and bacteria may trigger an inappropriate immune system response, leading to chronic inflammation in the intestinal walls and the digestive problems characteristic of Crohn disease. Researchers continue to study the relationship between changes in the *ATG16L1* gene and a person's risk of developing this disorder.

Chromosomal Location

Cytogenetic Location: 2q37.1, which is the long (q) arm of chromosome 2 at position 37.1

Molecular Location: base pairs 233,251,571 to 233,295,674 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- APG16 autophagy 16-like
- APG16L
- ATG16 autophagy related 16-like 1 (*S. cerevisiae*)
- ATG16 autophagy related 16-like protein 1
- ATG16A
- ATG16L
- Autophagy 16-like 1
- autophagy related 16-like 1
- autophagy related 16-like 1 (*S. cerevisiae*)
- WD repeat domain 30
- WDR30

Additional Information & Resources

Educational Resources

- Eurekah Bioscience Collection: Origin and Evolution of Self-Consumption: Autophagy
<https://www.ncbi.nlm.nih.gov/books/NBK6274/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ATG16L1%5BTIAB%5D%29+OR+%28APG16L%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- AUTOPHAGY 16-LIKE 1
<http://omim.org/entry/610767>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ATG16L1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ATG16L1%5Bgene%5D>
- HGNC Gene Family: Autophagy related
<http://www.genenames.org/cgi-bin/genefamilies/set/1022>
- HGNC Gene Family: WD repeat domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/362>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21498
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/55054>
- UniProt
<http://www.uniprot.org/uniprot/Q676U5>

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